

MISSOURI BIRTH DEFECTS

1996 – 2000

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Introduction

This report presents data on birth defects and other selected conditions among 1996-2000 Missouri resident live births, obtained from the Missouri birth defects registry. Birth defects, defined as abnormalities of structure, function, or biochemistry originating before birth, are a major but understudied contributor to infant mortality and morbidity. Approximately one in four Missouri infant deaths have a birth defect listed as cause of death. The true proportion is likely to be greater, as less than half of infant deaths are autopsied, and birth defects may be contributing factors in infant deaths with other listed causes.

While the genetic basis of some defects is known, and a few teratogens have been identified, the cause of most birth defects remains unknown and most cannot, at this time, be prevented. A notable exception is the ability to prevent 50-70 percent of neural tube defects (NTDs) with adequate folic acid intake before and during early pregnancy.¹⁻² Recent research indicates that increased folic acid intake may also reduce the risk for other birth defects, including oral clefts and congenital heart and urinary tract defects.³⁻⁴ (For further discussion of folic acid and NTDs, see pages 9-10).

The Missouri Department of Health and Senior Services (DHSS) established the birth defects registry to monitor the incidence of birth defects among Missouri infants, to provide data for epidemiological studies, surveillance for potential effects of environmental contamination, and to support and evaluate state programs serving ill and disabled children. Data are available for births since 1980, but improvements in the data set make data for births since 1993 much more complete than data for earlier years (Appendix A). In 1998, DHSS was awarded a cooperative agreement with the Centers for Disease Control and Prevention (CDC), which enabled DHSS to improve the timeliness of its data and to use registry data to improve access to state-provided services.

Data presented in this report are for liveborn Missouri residents. Defects must be diagnosed in the first year of life in order to be included in the registry. The registry is a data collection effort in which data available from a number of sources are merged. Data sources currently available are birth certificates, infant death certificates, newborn patient abstracts, pediatric patient inpatient and outpatient abstracts, the DHSS data base for children enrolled in programs such as Children with Special Health Care Needs, and the Department of Mental Health First Steps (early intervention) data base. Defects reported in each component are linked to the birth certificate to avoid duplication. The birth certificate also provides important demographic data not available in most of the individual components and provides a basis for producing rates and comparing attributes of infants with and without birth defects.

The Missouri birth defects registry is a passive data collection system; case finding is limited to reports provided to DHSS from the sources listed above, and verification of reported defects is not undertaken. It is probable that some Missouri infants have birth defects diagnosed in the first year of life that are not reported on any of the data components and are not, therefore, included in the registry; these are termed false-negative cases. Conversely, the registry will include cases termed false positive: spurious reports of birth defects that are the result of mis-diagnosis, mis-

coding, or tentative diagnoses that are later discounted. Some birth defects are not diagnosed in infancy and are therefore not included in the registry. Anomalies also affect pregnancies not resulting in live births. Birth defects often result in intrauterine death. The registry includes limited data on defects among spontaneous fetal deaths of 20 or more weeks gestation, but those data are not included in this report. Additionally, prenatally diagnosed defects often result in an elective termination of pregnancy. No registry data are available for elective terminations. Please read Appendix A for a further description of the birth defects registry and its components.

The purpose of this report is to provide descriptive statistics on the prevalence of birth defects in Missouri, by characteristics such as sex, race, maternal age, and area of residence; the association of birth defects with low birth weight, preterm delivery, and infant death; and participation of infants with defects in the Medicaid, Children with Special Health Care Needs, and First Steps programs.

Because of the complexities of the data, the reader is advised to read the technical notes and appendices in order to properly understand and use the statistics presented in this report.

Report Methodology and Terminology

The study population is 1996-2000 liveborn infants whose mothers resided in Missouri at delivery. It includes infants of Missouri residents born in other states.

Diagnostic Definitions

Defects are coded according to the International Classification of Disease-9th Revision (ICD-9). The ICD-9 codes comprising each diagnostic category presented in this report are listed in Appendix B. There is no standard definition of the specific conditions that should be included in a comprehensive, or overall, birth defect category. For this report, overall birth defect data refer to the number or rate of infants with one or more conditions classifiable to the ICD-9 congenital anomalies section (codes 740-759), excluding some insignificant anomalies listed in Appendix B.

Several other conditions not included in comprehensive birth defect statistics are listed at the bottom of tables detailing defects by type (Tables 2-6), under the heading “Other Selected Conditions.” These conditions are primarily of genetic or prenatal origin, but also include disorders that may arise postnatally, e.g., cerebral palsy and intracranial hemorrhage.

Rates, confidence intervals, and statistical significance

Unless otherwise specified, rates are per 10,000 live births. These statistics are sometimes based on a small number of cases and should be used cautiously. A rate is an estimate of the prevalence of a given condition. Exact binomial confidence intervals for overall birth defect rates by year of birth, race, sex, and maternal age are listed in Table 1. The 95 percent confidence interval represents a range of values within which we can state, with 95 percent confidence, the true rate lies. When the confidence intervals for two rates do not overlap, the observed difference in rates is considered statistically significant. Tables 2-4 and 7-9 include notations of statistically significant differences in rates, but not confidence intervals.

Methodology for determining statistical significance is that described above, i.e., calculation and comparison of 95 percent exact binomial confidence intervals.⁵

Race

Race-specific data are based on maternal race as listed on the birth certificate. Table 1 and Figure 2 include overall birth defect statistics for three race categories: white, black, and other. Defect-specific data for white and black infants are provided in Table 3; detailed data for other races are not provided because of the small number of defects for other race categories.

Maternal Age

Maternal age is that of the mother's age at delivery. Defect-specific data by age are presented in Table 4.

Low birth weight, preterm births, infant death

Data on two prematurity indicators are provided: low birth weight (LBW, less than 2,500 grams) and preterm (less than 37 weeks gestation). Of the two indicators, LBW is generally considered the more reliably reported, but because birth defects may affect birth weight independently of prematurity, both indicators are provided in this report. Infant deaths are those after birth and before the first birthday. Statistics are expressed as the number and percent of all LBW, preterm births and infant deaths that have one or more birth defect (Figure 5), or the number and percent of births with a defect that are LBW, preterm, and infant death (Table 5).

Medicaid

Medicaid statistics (Table 6) include the number and percent of births with defects to mothers who were Medicaid recipients during pregnancy, as listed on the birth certificate. Most infants of mothers enrolled in Medicaid are also enrolled at birth, and others not enrolled at birth may become Medicaid recipients later.

Children with Special Health Care Needs (CSHCN)

This DHSS program reimburses health care providers for many of the diagnostic and treatment services required for children with conditions including, but not limited to, many of the defects presented in this report. Eligibility is based on family income. While the CSHCN program provides assistance to children from birth to age 21, the statistics presented in this report (Table 6) are limited to the number and percent of infants enrolled in the first year of life.

First Steps

First Steps is an early intervention program for children, birth to age 3, who have delayed development or diagnosed conditions that are associated with developmental disabilities. First Steps is a Department of Elementary and Secondary Education program, and was developed in response to Part C of the Individuals with Disabilities Act. Among services provided are evaluation, counseling, and special instruction and therapy. There is no income requirement for participation in the program. For the years presented in this report, DHSS and the Department of Mental Health provided First Steps services and enrolled children in the program. Although the program offers services from birth to age three, statistics provided in this report (Table 6) are the number and percent of infants enrolled in the program before the first birthday.

County of residence

County of residence refers to the mother's residence at delivery. The number, rate, and statistical significance of the variation from the state are provided for four birth defect categories: overall birth defects (Table 7); neural tube defects (Table 8); oral clefts (Table 9); and Down syndrome (Table 10). Quartile rankings by county for these four categories are also displayed in Maps 1-4. If fewer than 80 percent of a county's births were linked to a newborn patient abstract, data are flagged and noted to be unreliable, and the county's rate is not included in quartile rankings. In Table 11, the frequency of selected birth defects is listed for each county.

County-specific data should be used with special caution. In addition to the problem of rates based on small numbers of events, other factors influence rates on a county level. Patient abstract data summarizing diagnoses made at birth and during subsequent hospitalizations are the most important source of birth defect data; some counties have a significant number of births in out-of-state or military hospitals for which patient abstract data were unavailable (see Appendix C). Additionally, differences in rates may only represent diagnostic coding practices prevalent in certain hospitals.

Data Highlights

Year of Birth

Overall birth defect rates ranged from 549 to 575 per 10,000 live births over the five-year period presented in this report (Figure 1). Of interest is the generally downward trend for neural tube defect rates from 8.1 per 10,000 in 1996 to 6.0 in 2000, with the lowest rate of 5.3 recorded in 1999 (Table 2). For a further discussion, see pages 9-10.

Figure 1
Birth Defect Rates by Year of Birth
Missouri Resident Data 1993-2000

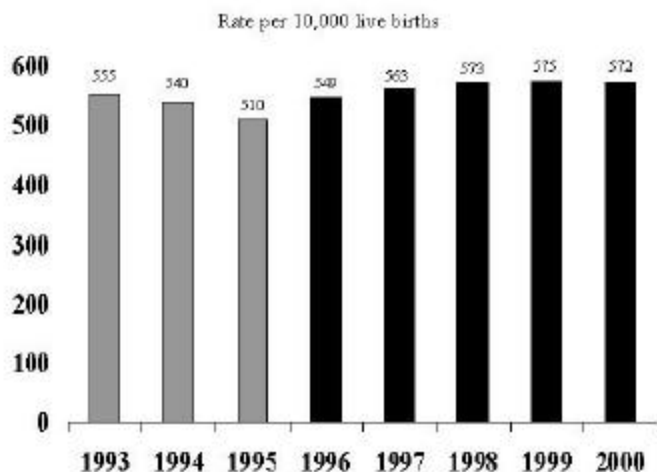
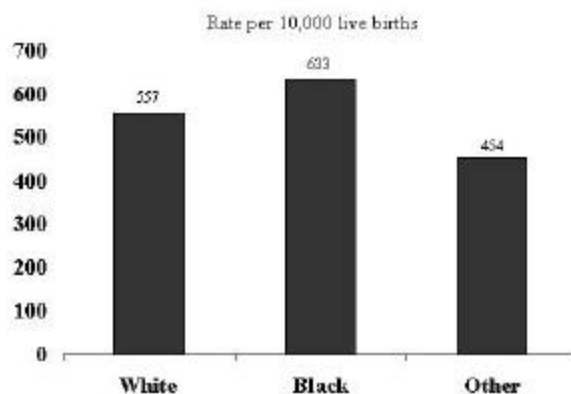


Figure 2
Birth Defect Rates by Race
Missouri Resident Data 1996-2000



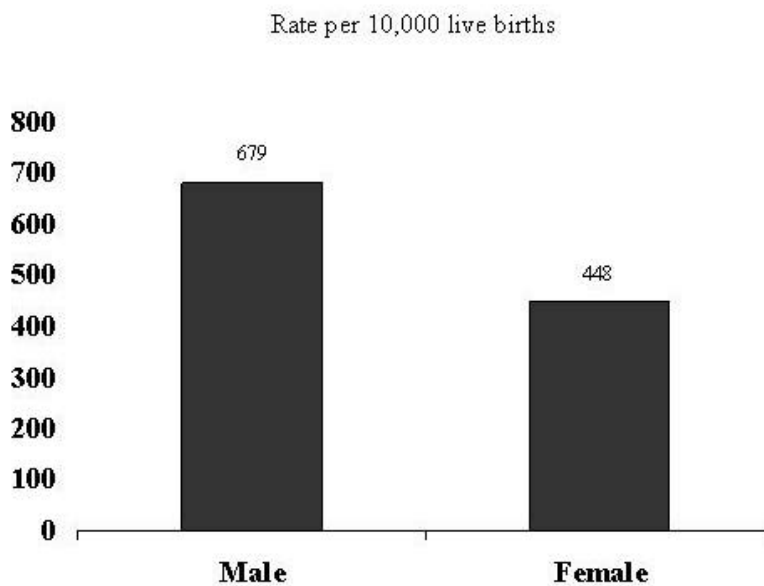
Race

The overall 1996-2000 birth defect rate is significantly higher for black infants than for white infants (Figure 2). The rate for other races was significantly lower than both the white and black rates; 70 percent of births in the other category were to Asian or Pacific Islander women. Among defects reported significantly more frequently for black infants were hydrocephalus, microcephalus, anomalies of eye, ear, face, and neck, heart and other circulatory anomalies, and integument anomalies (Table 3). White infants had significantly higher rates of oral clefts, digestive system anomalies, hip dislocations, and skull and facial bone anomalies. Among listed conditions not included in the overall birth defect statistics, congenital syphilis, sickle cell anemia, cerebral palsy, epilepsy, and intracranial hemorrhage were reported significantly more frequently for black infants.

Sex

For 1996-2000, male infants had a 52 percent higher overall birth defect rate than females (679 v. 448, respectively, Figure 3). Most of the difference in rates reflects higher rates of genital and urinary organ defects among male infants (Table 3), but male infants also had significantly higher rates for a number of other defect categories, including hydrocephalus, heart and other circulatory/respiratory anomalies, cleft lip with or without cleft palate, pyloric stenosis, clubfoot, and skull and facial bone anomalies. Female infants had higher rates of microcephalus and congenital hip dislocation.

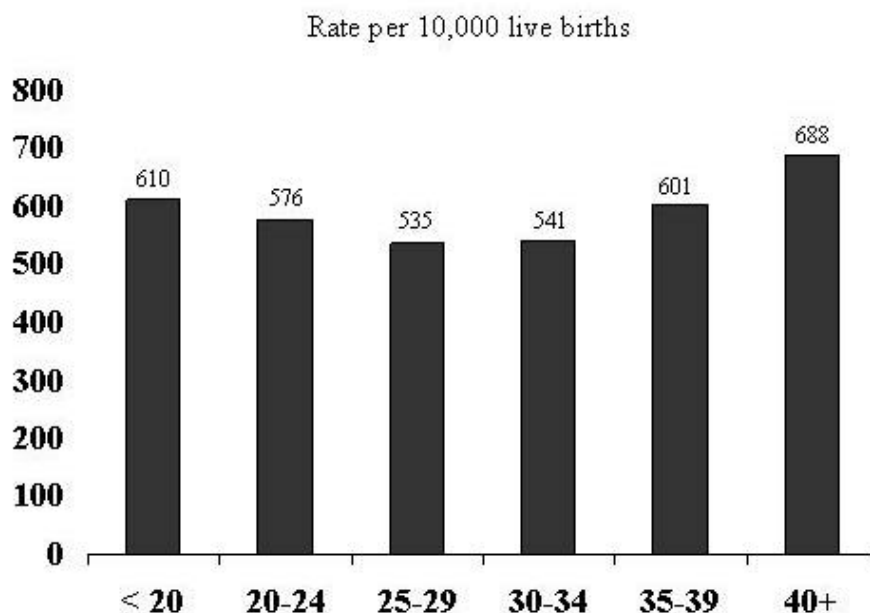
Figure 3
Birth Defect Rates by Sex
Missouri Resident Data 1996-2000



Maternal age

The overall risk of birth defects is increased at both ends of the maternal age spectrum (Table 4, Figure 4), highest for infants of mothers aged 40 plus, but also elevated for mothers less than 25. Chromosomal anomalies account for most of the increased risk for women aged 35 plus, but infants of older mothers also had significantly higher rates of heart and other circulatory/respiratory defects, atresia or stenosis of the small intestine, and urinary organ anomalies. Compared to infants of mothers aged 25-34 years, infants of both younger and older mothers were at higher risk of central nervous system defects. Infants of women less than 25 also had higher rates of atrial septal defects, digestive system, and musculoskeletal and integument defects. Additionally, they were at higher risk for congenital pneumonia and intracranial hemorrhage.

Figure 4
Birth Defect Rates by Maternal Age
Missouri Resident Data 1996-2000



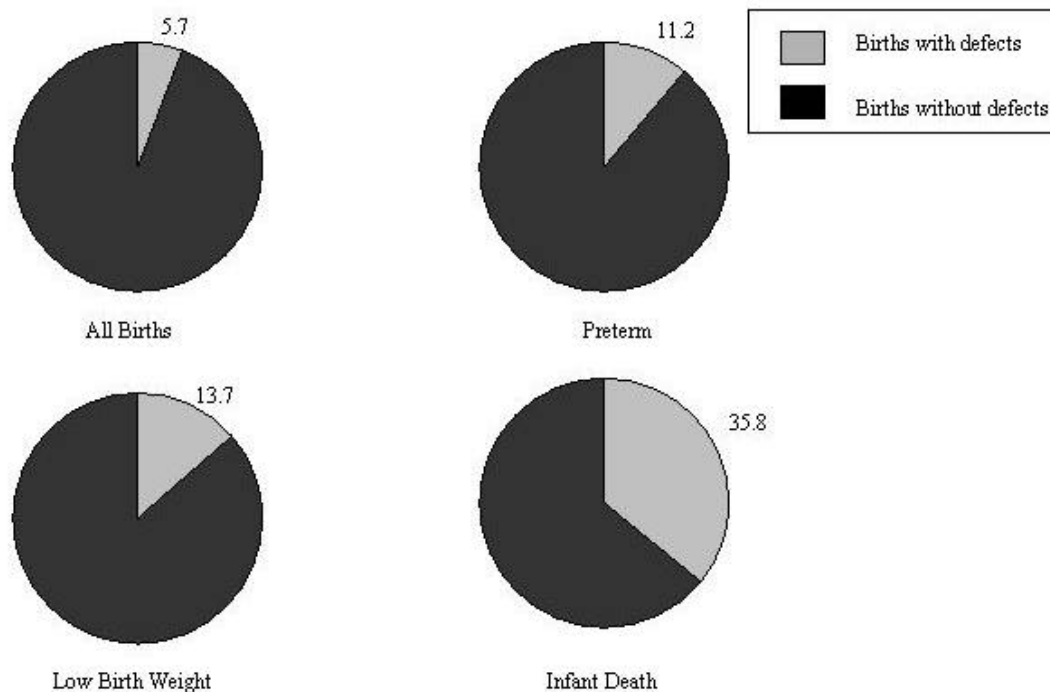
Low Birth Weight and Preterm Births, Infant Death

As shown in Figure 5, while births with defects represented only 5.7 percent of all births, they represented 11.2 percent of preterm births, 13.7 percent of low birth weight (LBW) and 35.8 percent of infant deaths (including infants with defects whose listed cause was not a birth defect).

The number and percent of births with defects that are LBW, preterm, and infant deaths are listed in Table 5. Overall, births with defects had an 18.7 percent LBW rate and a 20.1 percent preterm rate, in comparison with LBW and preterm rates of 6.7 and 9.1 percent, respectively, for infants without defects. Most defect categories had elevated rates for both indicators. In general, LBW and preterm rates are also high among infants with the other conditions presented but not included in birth defect statistics, particularly intracranial hemorrhage, because prematurity is a major risk factor for development of this condition.

In addition to anencephaly, an invariably fatal condition, defects with infant death rates in excess of 60 percent are hypoplastic left heart syndrome, agenesis of the lung, and trisomies 13 and 18. Overall, 4.8 percent of births with defects died in infancy, compared to 0.5 percent of infants without birth defects noted.

Figure 5
Percent of Total Births, Preterm and Low-Birth Weight Births, and
Infant Deaths with One or More Birth Defects
Missouri Resident Data 1996-2000



Medicaid, CSHCN and First Steps Participation

The number and percent of births participating in the Medicaid, Children with Special Health Care Needs (CSHCN) and First Steps programs are listed in Table 6. The 44.1 percent rate of Medicaid participation at birth among all infants with defects is slightly higher than the 40.5 percent rate for all births. Among infants with gastroschisis and other abdominal wall defects, 63.7 percent were Medicaid participants, reflecting the preponderance of gastroschisis among infants of poor and young mothers.

In the first year of life, 5.1 percent of infants with defects were enrolled in the CSHCN program, including 25.9 percent of infants with spina bifida and 22.9 percent of those with hydrocephalus. The program also served 23.2 percent of infants with cerebral palsy.

The First Steps program enrolled 11.0 percent of infants with defects in the first year of life. These include 57.6 percent of infants with spina bifida, 54.3 percent of those with hydrocephalus, and 68.6 percent of Down syndrome infants. In addition, 83.2 percent of infants with infantile cerebral palsy and 60.8 percent of infants with epilepsy were enrolled as infants in the First Steps program.

County of Residence

Overall birth defect rates by county of residence are listed in Table 7, and pictured in Map 1. Counties with an overall birth defect rate in the highest quartile and significantly higher than the rest of Missouri were Adair, Camden, Cape Girardeau, Christian, Cole, Dent, Phelps, and Stone. Although the overall birth defect rates for Franklin and Greene counties were significantly high in comparison with the state rate, the rates for these counties were not in the top 25 percent of overall birth defect rates by county. Caution should be used when evaluating differences in rates between counties, especially the overall birth defect rate. This category includes many diagnoses, including relatively minor defects, and differences between counties may largely reflect differences in reporting practices among the hospitals utilized by a county's patients.

Neural tube defects are distributed relatively evenly throughout the state (Table 8, Map 2). Only Osage County's NTD rate was significantly higher than the state rate. The counties making up the major metropolitan (St. Louis and Kansas City) areas have a somewhat lower NTD rate than the rest of Missouri (6.1 v. 6.8, respectively), but the difference is not statistically significant.

Oral cleft rates are significantly higher in the rural counties (Table 9, Map 3). Oral clefts are somewhat more common in white than black infants (Table 3), and rural Missouri counties have a much higher proportion of white infants. However, the racial make-up of the two groups only explains a portion of the difference in oral cleft rates between metropolitan and non-metropolitan counties. The rates for both white and nonwhite infants in nonmetropolitan counties are higher than the corresponding metropolitan rates. Knox, Scott, and Wayne counties had significantly elevated oral cleft rates.

Down syndrome rates by county are shown in Table 10 and Map 4. No county had a significantly elevated rate of Down syndrome. Metropolitan and non-metropolitan rates are similar.

Neural Tube Defects and Folic Acid

The neural tube is an early embryonic structure that develops into the brain, spinal cord, and supporting bone structures. Neural tube defects (NTDs) occur when the neural tube fails to develop properly. These defects occur very early in pregnancy, between the 17th and 30th days after conception, that is, before most women know that they are pregnant.

The two major NTDs are anencephaly and spina bifida. Anencephaly is a fatal condition in which the brain is absent or fails to develop completely. Anencephalic pregnancies often result in fetal death, and liveborn anencephalic infants die soon after birth.

Spina bifida occurs when the lower end of the neural tube fails to close, resulting in improper development of the spinal cord and backbones. A sac containing spinal fluid, and in some cases a portion of the spinal cord, may protrude from the back. Most babies born with spina bifida survive infancy; the 1996-2000 Missouri survival rate was 93 percent. Surviving infants face varying degrees of morbidity and disability, including paralysis of the legs, hydrocephalus, bladder and bowel control problems, and learning disabilities.

In 1992 the Center for Disease Control and Prevention (CDC) concluded that 50-70 percent of NTDs could be prevented by daily consumption of 400 mcg (0.4 mg) of folic acid before and during early pregnancy.⁶ Folate is one of the B vitamins and occurs naturally in many foods, including eggs, beans and peas, oranges, and many green vegetables, but it is difficult for women to consume the recommended amount through diet alone. Most multi-vitamins contain the recommended amount of folic acid. The Food and Drug Administration authorized optional folic acid fortification of enriched grain products beginning March 1996 and mandatory fortification beginning January 1998. Fortification was expected to add 100 mcg of folic acid to the average daily diet.⁷⁻⁸

NTDs in Missouri

Each year in Missouri, about 10 infants are born with anencephaly and 38 with spina bifida. The number of affected pregnancies is considerably higher. Each year NTDs are noted on approximately 8 fetal death certificates (completed for pregnancies of 20 or more weeks gestation). Other NTD-affected pregnancies result in earlier fetal loss for which no data are available. Many NTD cases are detected prenatally and are electively terminated. Although the DHSS birth defects registry does not include data for elective terminations, we were able to obtain the number of 1997-99 NTD-affected elective terminations from five major abortion providers. For those years, the facilities collectively averaged 9 terminations per year of NTD-affected pregnancies.

The rates of spina bifida among 1993-2000 Missouri live births are displayed in Figure 6, and those for anencephaly in Figure 7. The 1997-2000 spina bifida rate of 4.6 per 10,000 is 32 percent lower than the 6.8 per 10,000 rate for 1993-96. The decline in the spina bifida rate in Missouri is temporally associated with folic acid grain fortification, although other factors may be associated with the decrease. Anencephalus did not have a similar decline. Between 1993 and 2000, the rates for anencephaly ranged from 0.7 to 2.9 per 10,000, with no temporal trend.

Similar trends in NTDs have been observed elsewhere. Birth certificate reports of NTDs in 45 states revealed that from 1995 to 1999, there was a statistically significant 23 percent decline in reported cases of spina bifida, with an 11 percent decline in anencephalus that was not statistically significant.⁹

Figure 6
Spina Bifida Rates per 10,000 Live Births
Missouri Resident Data 1993-2000

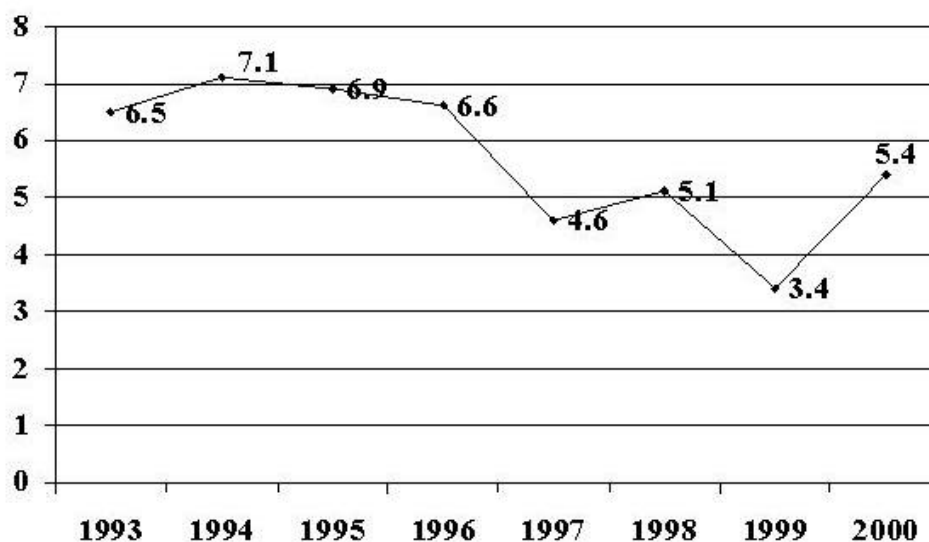
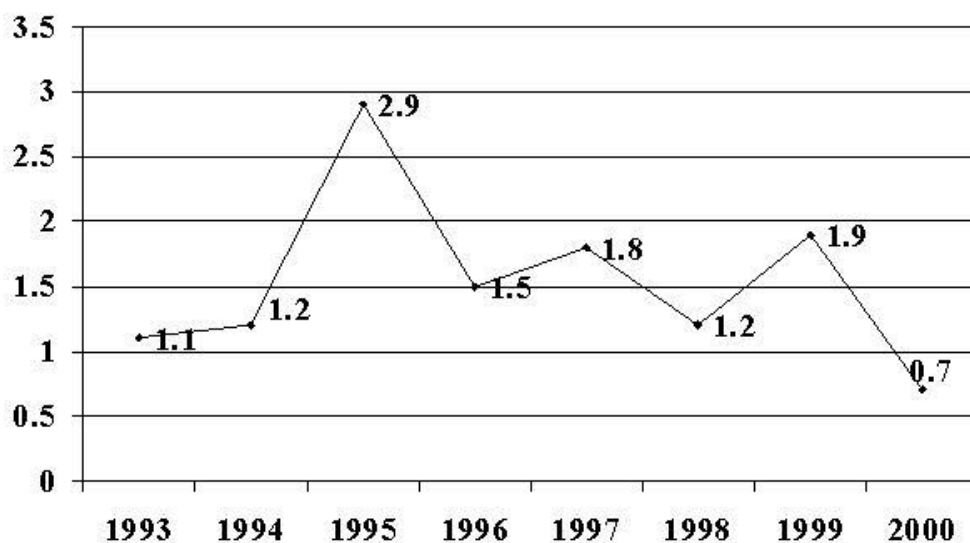


Figure 7
Anencephaly Rates per 10,000 Live Births
Missouri Resident Data 1993-2000



Recommendation

Neural tube defects are among the most serious of birth defects, and the identification of the effectiveness of adequate folic acid intake in preventing NTDs is one of the most promising public health developments in recent years. Significant declines in the birth prevalence rate of spina bifida, but not anencephalus, have been observed in Missouri and other states, and may be associated with folic acid fortification of grain products. However, the level of folic acid fortification of grain products does not result in the consumption by most women of the recommended daily intake of folic acid. The U.S. Public Health Service strongly recommends that all women capable of becoming pregnant—not just those planning a pregnancy—should consume 400 mcg (0.4 mg) of folic acid daily.⁶ A multi-vitamin containing folic acid is an effective method of obtaining the recommended daily intake. Folic acid supplementation is urged for all women capable of childbearing because NTDs occur too early in pregnancy to prevent after a pregnancy is recognized, and because half of all pregnancies are unplanned. Future research may establish the effectiveness of folic acid in reducing the risk of other types of birth defects²⁻⁴, giving additional emphasis to the importance of folic acid for healthy pregnancies.

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